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# Genetic Counselling and Testing for Women's Cancers





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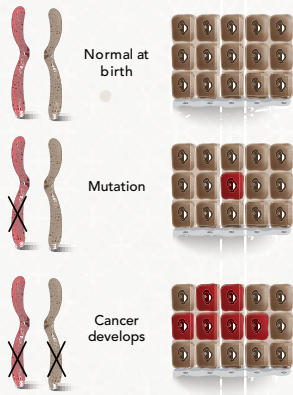
# Overview of Cancer



Cancer is a common disease. The chances of cancer increase with age. It is a multifactorial disease resulting from the combined influence of genetic changes and environmental factors.<sup>1</sup>

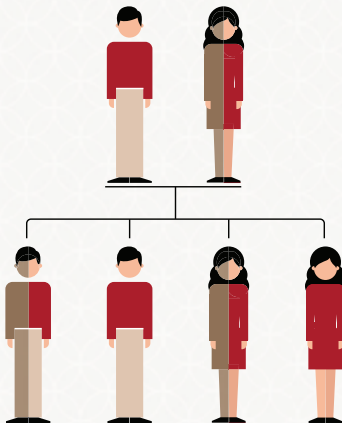


# Overview of Cancer



Over **75% of cancer patients have sporadic cancer**, which is mainly caused by age and environmental factors.<sup>2</sup>

About **15% – 25%** of cancer patients have a family history of cancer but do not have a specific genetic mutation identified. It is known as **familial cancer**.<sup>1,3</sup>

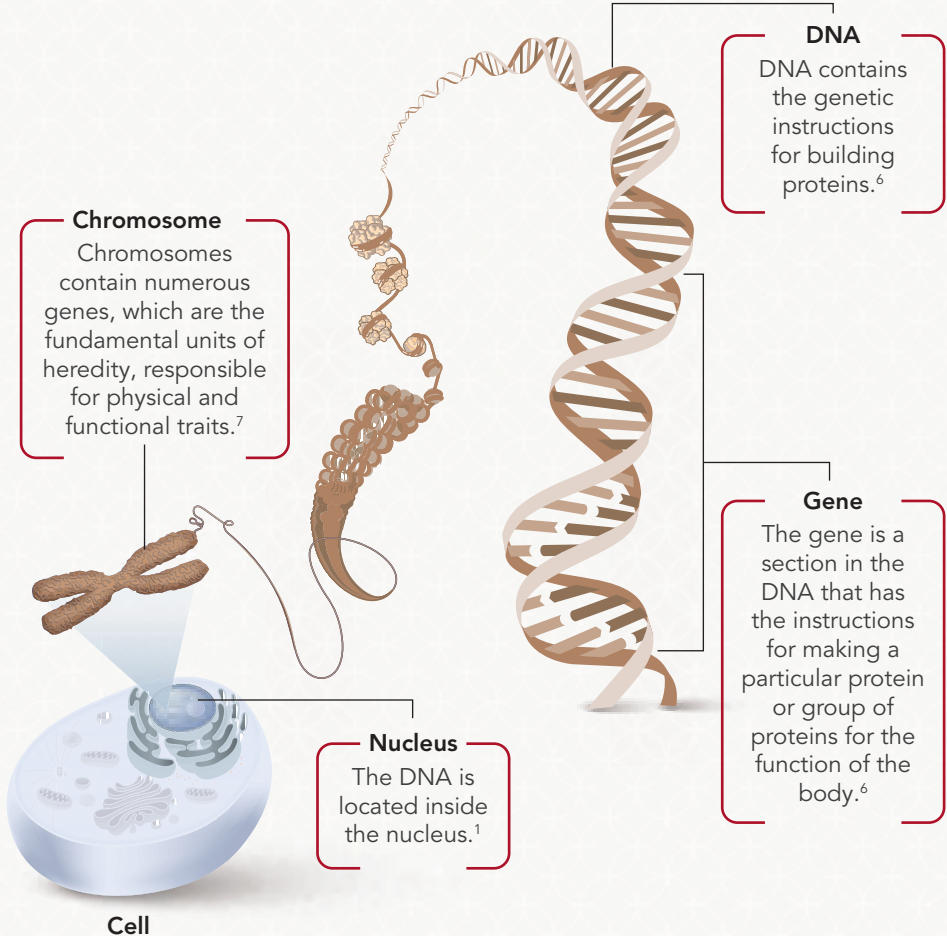


**Hereditary cancer affects 5% – 10%** of patients who are **born with a genetic mutation**. This increases their lifetime risk of developing certain cancers and can be passed from one generation to another.<sup>4,5</sup>

# What Are Genes?



Genes are the instructions encoded in our deoxyribonucleic acid (DNA) that dictate the way the body functions. We have over **25,000 genes** in our body.<sup>6</sup>



“All genes, except those on sex chromosomes, come in pairs as we inherit one copy from our mother and the other copy from our father.”<sup>8</sup>

# What Are Genetic Mutations?



Genetic mutations are changes in DNA similar to typographical errors that can provide wrong instructions, leading to abnormal functions in our bodies.<sup>1</sup>



**DNA changes that cause diseases are called 'genetic mutations'.<sup>1</sup>**

# Types of Genetic Mutations

Genetic mutations can either be acquired (referred to as 'somatic') or inherited (referred to as 'germline').<sup>8</sup>

## Somatic DNA changes

Acquired over a person's lifetime in single cells



Can lead to cancer

Cannot be inherited

## Germline DNA changes

Present in every cell of the body, including eggs and sperm



Can be inherited

Can increase risk of developing cancer

Examples: Hereditary breast and ovarian cancer syndrome and Lynch syndrome<sup>7,9</sup>

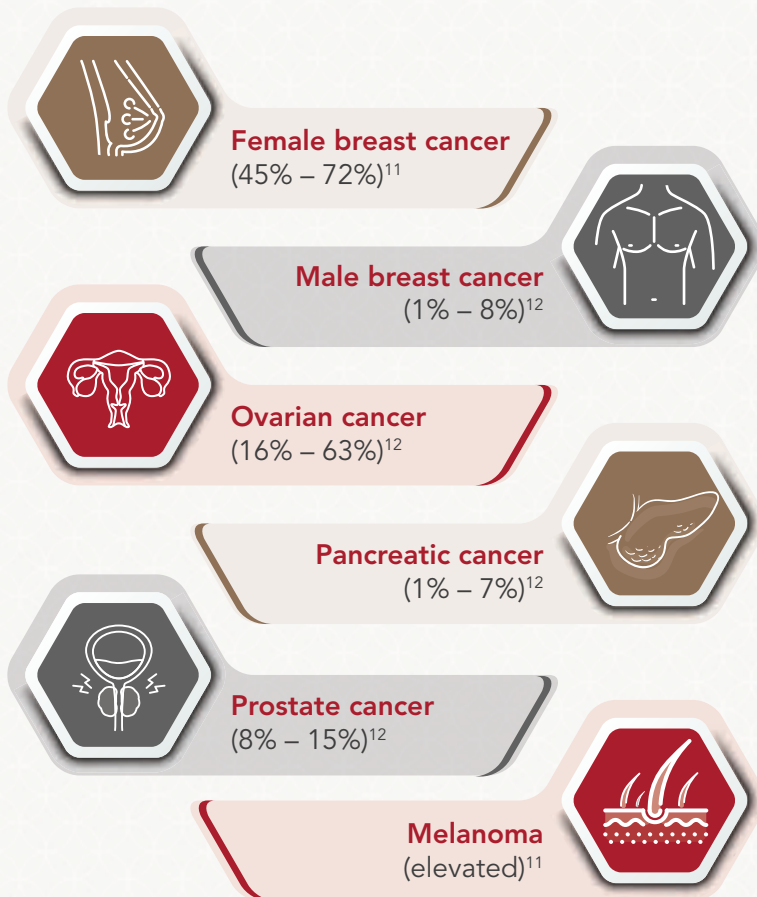


# What Is Hereditary Breast and Ovarian Cancer (HBOC)?



**BR**east **CA**ncer gene 1 (**BRCA1**) or **BR**east **CA**ncer gene 2 (**BRCA2**) genetic mutations are two of the most common mutations associated with **H**ereditary **B**reast and **O**varian **C**ancer (**HBOC**).<sup>10</sup>

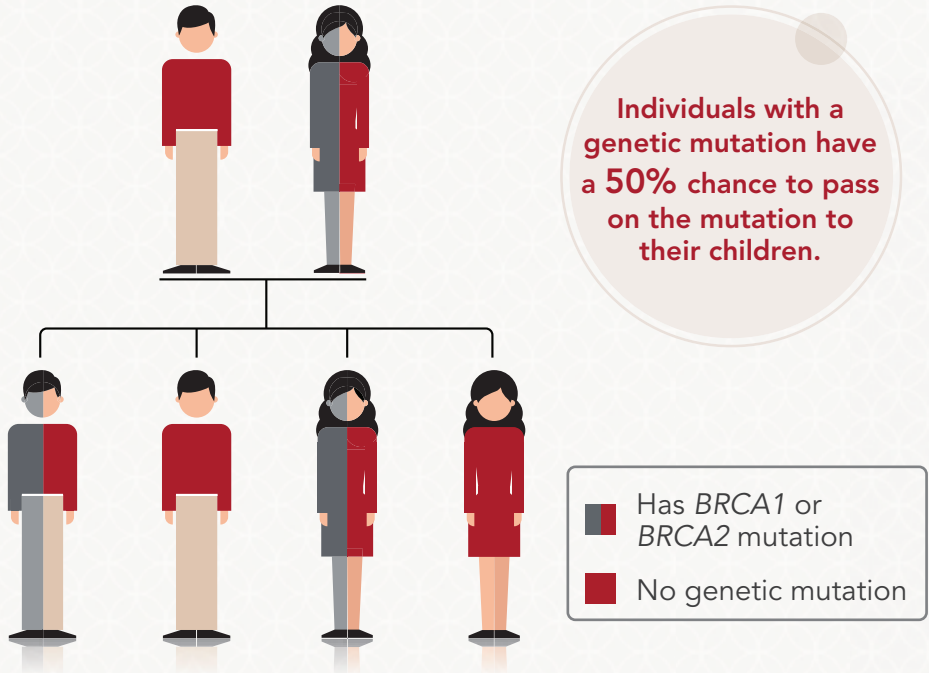
The **BRCA1** and **BRCA2** genetic mutations are linked to an increased risk of developing several types of cancer, including:



# How Is HBOC Inherited?



In HBOC, the inherited genetic mutation, such as the *BRCA1* or *BRCA2* gene, is present in all the cells of an individual and passed down from a parent (father or mother) to the child.<sup>13,14</sup>



Not everyone who inherits the genetic mutation will develop cancer in their lifetime.<sup>13</sup>

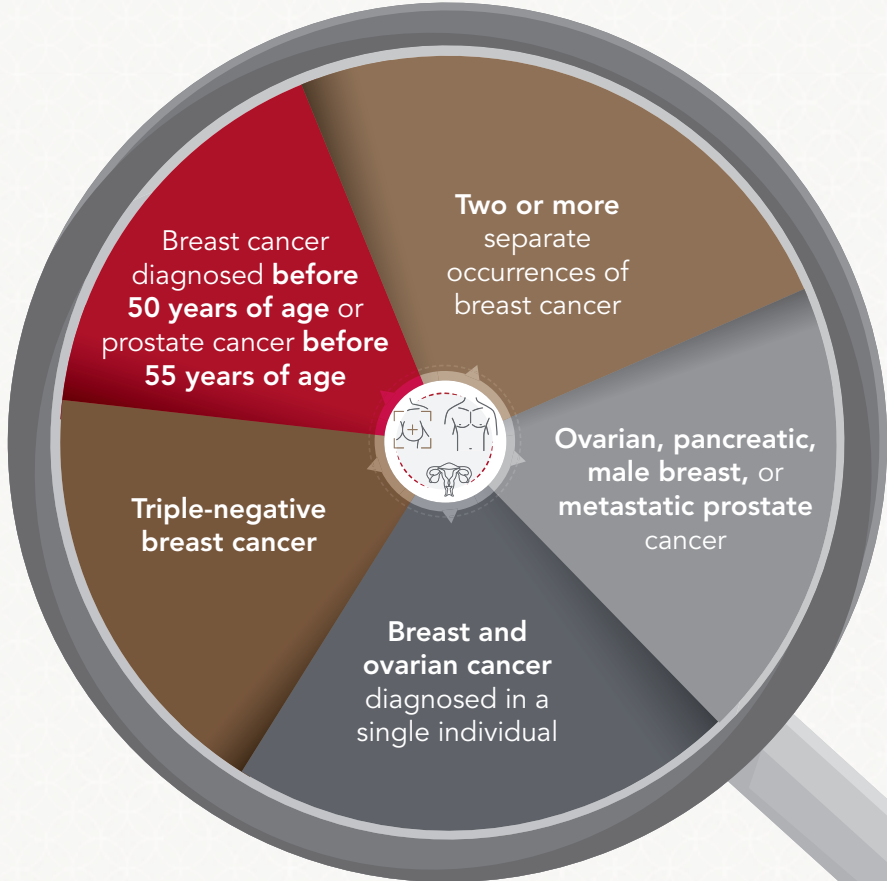


If an individual is found to carry a genetic mutation that can cause HBOC, his or her immediate family members may have an increased risk of inheriting the same genetic mutation. To verify this, a **genetic test can be conducted.**<sup>13</sup>

# Who Should Consider Genetic Testing?



Individuals with a personal or family history of:<sup>11</sup>



# What Is Genetic Testing?



Genetic testing is conducted to help individuals **make informed decisions about cancer treatment or prevention**. It also **provides information about potential implications that may affect their family**.<sup>16</sup>

**Two types of genetic testing** can be performed:<sup>17</sup>

## Tumour Test

For individuals who have been diagnosed with cancer and to guide their treatment options<sup>17</sup>

Tumour test sample taken via:<sup>17</sup>



Tumour biopsy



Liquid biopsy

## Germline Test

To identify specific inherited genetic mutations that have been linked to an increased risk of certain cancers<sup>17</sup>

Germline test sample taken via:<sup>17</sup>



Blood



Saliva



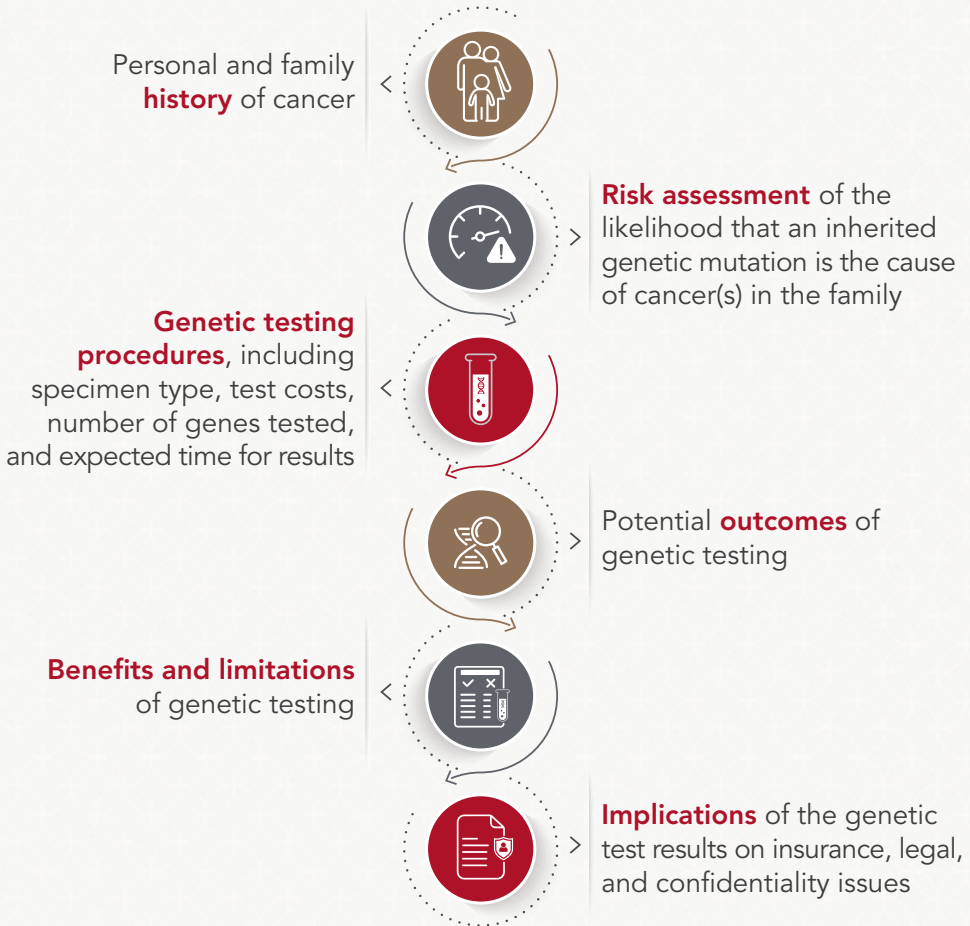
Skin

# What Is the Process of Genetic Counselling?



Genetic counselling is **conducted before an individual undergoes germline genetic testing**. This process provides patients and/or their family members with information to help them make informed decisions about cancer risk, treatment of cancer, and potential implications for their family.<sup>18,19</sup>

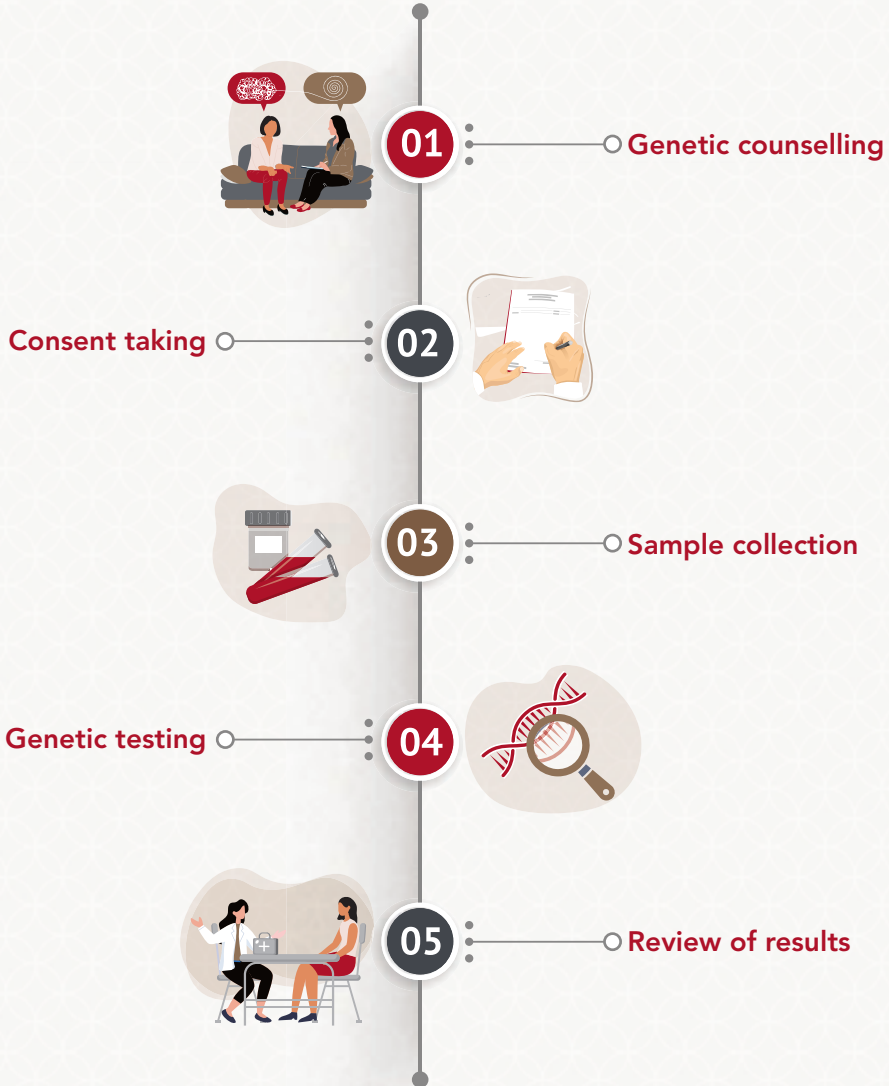
**Topics that will be covered during a genetic counselling session include:**<sup>18,19</sup>



# What Is the Procedure for Genetic Testing?



Genetic testing is a personal choice. If an individual agrees to undertake germline genetic testing, written informed consent is required. Blood or saliva samples will be collected for testing, and results will usually be ready in a few weeks.<sup>20</sup>



# What Are the Potential Genetic Test Results?



Genetic testing for an inherited mutation can have **three possible results**.<sup>18,20</sup>



## Positive

- A genetic mutation known to cause hereditary cancer is identified.
- Medical management, such as increased surveillance or risk-reducing surgery, may be recommended.
- Predictive testing is recommended for immediate family members.



## Negative

- No genetic mutation known to cause hereditary cancer found.
- This does not rule out a genetic cause for cancer.



## Variant of uncertain significance

- An uncertain gene change(s) is identified. However, it is unclear if the change(s) increases your risk of cancer.
- In some instances, this result may be clarified by testing other family members and could be reclassified over time as 'positive' or 'negative'.
- Medical management should be based on personal and/or family history.



# What Are the Benefits of Germline Genetic Testing?

Provides an explanation for your personal or family history of cancer<sup>18</sup>



Determines your risk of cancer to help you make informed medical decisions, such as regular screening to detect cancer as early as possible and/or risk-reducing surgery<sup>18</sup>



Identifies other family members who may be at risk of developing cancer<sup>18</sup>



Guides treatment options for current cancer diagnosis<sup>18</sup>



If you have a personal or family history of cancer or are interested in learning about your inherited cancer risk, **talk to your doctor about whether germline genetic testing is right for you.**



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# Notes



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